Aspects of Sexual Medicine

Genetic Counselling

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Until the early 1960s little genetic counselling was done, and few people appreciated its value or even its need. But the situation has changed and nowadays almost every medical school and teaching hospital supports a genetic counselling unit. Before considering the problems of genetic counselling perhaps we should first be clear exactly what we mean by "genetic disease."

There are essentially three categories of genetic disease. Firstly there are the *unifactorial* disorders, each of which is due to a single (Mendelian) gene which may be dominant, recessive, or X-linked. Examples are given in table I. These disorders are individually rare but the risks to relatives are usually high. Secondly, there are the *chromosomal* disorders such as Down's syndrome (mongolism) and certain disorders associated with male infertility (Klinefelter's syndrome) or primary amenorrhoea (Turner's syndrome). Thirdly, there are the *multifactorial* disorders which result from the effects of many genes plus environmental effects. They include many congenital malformations (anencephaly, spina bifida, hare-lip, and cleft palate), diseases of "modern society" (hypertension, coronary artery disease, peptic ulcer, diabetes mellitus), and certain psychiatric disorders (schizophrenia and probably manic-depressive psychosis).

Extent of Problem

With advances in medicine and surgery and the concomitant decline in infectious diseases and nutritional deficiencies, the proportion of morbidity and mortality due to genetic disease has increased. In fact at present roughly 1 in 20 children admitted to hospital have a unifactorial or chromosomal disorder, and such disorders account for about 1 in 10 of childhood deaths. In contrast only about 1 in 100 adult inpatients has a unifactorial or chromosomal disorder, but then many of these disorders lead to early death, or if they are compatible with survival to adulthood they usually do not warrant hospital admission.

The prevention of genetic disorders depends on ascertaining those individuals in the population who are at risk of having affected children and providing them with genetic counselling. Unfortunately only a relatively small proportion of those at risk are referred for genetic counselling. Some might argue that the ascertainment of those at risk should not be left to chance. For this reason it has been suggested that a confidential, computerized register of families with genetic disorders might prove valuable as a means of ascertaining and following up individuals at risk. Such a Register has been started in Edinburgh under the acronym RAPID (*R*egister for the *A*scertainment and *P*revention of *I*nherited *D*isease).

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Changing Patterns in Genetic Counselling

Whereas at one time the majority of people seen for genetic counselling were married and in the higher social classes and were usually referred by a consultant interested in the disorder in question, this is no longer so. Recent follow-up studies indicate that an increasing proportion are referred by family doctors, and increasingly requests come from individuals themselves, often as a result of articles they have read or programmes they have seen on television. There is no longer a preponderance of professional couples, and increasingly more individuals seek advice before marriage. It is clear therefore that an awareness of genetic disease and the importance of genetic counselling is extending into the general population.

GENETIC RISKS

In attempting to define the risk of recurrence the first prerogative is a precise diagnosis. When the diagnosis is well established and the mode of inheritance is clear, genetic counselling is straightforward. However, a serious complication is the existence of genetic heterogeneity. This term refers to disorders which are clinically similar but are inherited in different ways, and the individual's family history often gives no clue to this. In such cases the patient is best advised by a specialist genetic counsellor. In unifactorial disorders the risks of recurrence follow Mendelian principles (table I). The risks in the commoner multifactorial disorders can be derived from data on the prevalence of the disorder in relatives of affected individuals (table II).

TABLE I—*Examples of Unifactorial Disorders*

Dominant Disorders: Risk to offspring of affected individuals is 1 in 2	
Achondroplasia (classical)	Osteogenesis imperfecta
Huntington chorea	Polyposis coli
Marfan syndrome	Tuberous sclerosis (epiloia)
Neurofibromatosis	Polycystic kidney disease of adults
Recessive Disorders: Risk to further children of healthy parents is 1 in 4; risk to offspring of affected individuals is usually negligible	
Fibrocystic disease	Phenylketonuria
Friedreich ataxia	Sickle-cell anaemia
Galactosaemia	Tay-Sachs disease
Glycogen storage diseases	Thalassaemia
Homocystinuria	Werdnig-Hoffmann disease
Hurler syndrome	Wilson disease
X-Linked Disorders: Risk to male offspring of healthy carrier females is 1 in 2 (there is a 1 in 2 chance that a daughter will also be a carrier)	
Duchenne muscular dystrophy	
Glucose-6-phosphate dehydrogenase deficiency	
Haemophilia	

In Down's syndrome the risks of recurrence depend on the cause. Most cases are due to an extra chromosome 21 (trisomy 21). In these cases the risks of recurrence in future children may be as high as 1 in 100 in women who have previously had an affected child, and 1 in 50 in women over the age of 40. Occasionally (no more than 3% of cases) the disorder is due to an inherited chromosomal translocation, in which case the chances of recurrence are greater than 1 in 20 depending on the type of

translocation and whether the mother or the father is the translocation carrier. It is therefore important in all cases of Down's syndrome to check first the child's chromosomes and, if a translocation is found, to check the parents' chromosomes.

TABLE II—*Approximate Empirical Risks of Recurrence for some Common Disorders (Modified from Emery[7])*

Disorder	Normal Parents having a *Second* Affected Child	Affected Parent having an Affected Child
Anencephaly + spina bifida	1 in 20	1 in 20 (parent with spina bifida)
Cleft palate only	1 in 50	1 in 14
Cleft lip ± cleft palate	1 in 25	1 in 25
Congenital heart disease (all types)	1 in 30	1 in 30
Diabetes mellitus:		
early onset	1 in 12	1 in 12
late onset	—	1 in 10
Epilepsy ("idiopathic")	1 in 20	1 in 20
Manic-depressive psychosis	—	1 in 7
Mental handicap ("non-specific")	1 in 20	—
Profound childhood deafness ("idiopathic")	1 in 10	1 in 16
Schizophrenia	—	1 in 7

PHILOSOPHY OF GENETIC COUNSELLING

At the outset it must be emphasized that parents should never be told what to do. Ideally they should be provided with all the information, within the framework of their educational background, necessary to help them arrive at an informed decision. When the diagnosis of a serious genetic disorder is first made is not the time to give genetic counselling, as parents are often upset and confused. They are more likely to welcome advice later on. Whenever possible genetic counselling should be given when both parents can be present at the same time.

The first step is to remove the parents' feelings of guilt and self-recrimination which often accompany the realization that a child has a genetic disorder. Next, the nature of the disease itself should be discussed and, in the simplest of terms, what is meant by saying that it is "genetic." The prognosis and the availability of treatment need to be made clear. Finally the risks of recurrence are presented, again within the framework of the parents' educational background. Parents often find mathematical probabilities difficult to comprehend, and in such cases it is better to emphasize the risks in less precise terms.

If a person is found to be at high risk (usually defined as greater than 1 in 10) of having an affected child, various possibilities may have to be discussed, such as family limitation, sterilization, and antenatal diagnosis.

A problem which often arises is whether parents should be told that they are at risk of having an affected child if they have not requested this information—for example, after the diagnosis of Huntington's chorea in the father or mother of a would-be parent. I feel that parents have a right to know these risks if it might prevent the birth of an affected child. A doctor who decides to withhold such information from a family assumes a heavy responsibility. The family doctor is a good guardian of the individual's interest in this regard. I have found that in situations like this it is best to discuss the genetic risks and their implications with the family doctor in the first instance.

VALUE OF GENETIC COUNSELLING

Since most genetic counselling is provided only *after* the birth of an affected child, because only then is the need in any particular family recognized, it can prevent only some cases of genetic disease. For example, the proportion of cases which could be prevented by genetic counselling is about 20% in the case of recessive and severe X-linked disorders and at most 5% in the case of multifactorial and chromosomal disorders. In dominant disorders it depends on the fertility of affected individuals. The closer this is to normal the greater the proportion which might be prevented by genetic counselling in families with an affected individual. If parents were screened *before* having children, a much greater proportion of genetic disease could be prevented, but in fact at present this is scientifically and economically possible only in a very few cases—for example, Tay-Sachs disease in Ashkenazi Jews. However, though these figures indicate that genetic counselling might not have a profound effect on the frequencies of genetic disorders in the population, this completely ignores the value to the individual family in providing reassurance for those who prove not to be at risk and in presenting various possibilities, such as antenatal diagnosis in the fetus, to those found to be at high risk (table III).

TABLE III—*Main Indications for Antenatal Diagnosis at Present*

Cytogenetic studies
- (*a*) Down's syndrome: Maternal age >40; familial chromosomal translocation; previous trisomy-21 child
- (*b*) X-linked recessive disorders: Sexing of fetus (for example, Duchenne muscular dystrophy and haemophilia)

Biochemical studies
- (*a*) Recessive and X-linked disorders (for example, Tay-Sachs disease, galactosaemia, Lesch-Nyhan syndrome)
- (*b*) C.N.S. malformations (anencephaly and spina bifida)

RESPONSE TO GENETIC COUNSELLING

The individual's response to genetic counselling depends on the severity of the disorder, the availability of effective treatment, the risks of recurrence, religious attitudes, and socio-economic factors, probably in this order.

When faced with a high risk of having a child with a serious genetic disorder experience shows that many parents accept the risk and plan future pregnancies if the disorder is very severe and likely to be a "burden" for only a limited period. An example is Werdnig-Hoffmann disease, or progressive spinal muscular atrophy. In other situations the possibilities open to a couple are family limitation with therapeutic abortion if this fails, sterilization of one of the partners, artificial insemination by donor (A.I.D.) if both parents carry the same rare recessive gene or if the husband has a dominant disorder, and antenatal diagnosis.

In our studies we have been disturbed to find that contraceptive measures failed in as many as 1 in 10 couples who wished to avoid further pregnancies because of the risks involved. There is also no doubt that the fear of having another affected child has seriously impaired marital harmony in some families we have studied, with resulting separation and divorce. For these reasons if a couple is at high risk of having a child with a serious genetic disorder and therefore they do not wish to have further children, expert contraceptive advice should be provided. Not only this; couples must be given ample opportunities to discuss related anxieties, perhaps the most important of which will involve their marital relationships. Such problems may not be obvious at a casual interview—for example, in a busy outpatient clinic—and the physician may have to ask directly about such matters. In our experience these problems have sometimes come to light only when a social worker has made a home visit. Sterilization may be considered the best answer in some cases, but a little caution is required because increasing numbers of disorders can now be diagnosed in utero in early pregnancy, and if the fetus is found to be effected the parents can be offered selective abortion. In any disorder which cannot yet be diagnosed in utero this possibility at some time in the future has to be balanced against the risks of pregnancy in the intervening period. Adoption is no longer an obvious answer because of the decreasing number of babies which are available for adoption.

Role of the Family Doctor

The help and advice of a family doctor are of great assistance in genetic counselling. In straightforward cases he is best

equipped to give such counselling himself. He can help the parents make their decision, which the specialist may find difficult because he usually has little idea of a couple's economic and social background. Our follow-up studies have repeatedly shown that genetic counselling in many cases is best given, or at least reinforced, in the home environment. Even with more complex problems, in which the geneticist has been asked to determine the risks—perhaps having to base such calculations on the results of special tests on the parents—the role of the family doctor is important. He can reinforce the advice given by the geneticist and probably enable the parents to understand it better.

Management of a Family with a Genetic Disorder

The first step in the management of a family with a genetic disorder is to establish the precise diagnosis. This may mean soliciting the advice of a hospital specialist and having access to death certificates and pathology reports. Secondly, the risks of recurrence have to be established. Here several publications can be helpful.[1-5] If expert advice is required, it may be obtained from one of the genetic advisory centres listed in a publication entitled "Human Genetics" from the Health Department.[6] In many straightforward cases there is no doubt that genetic advice can be and is perhaps best given by the family doctor. In giving such advice, however, all doctors should bear in mind the complications, such as genetic heterogeneity, that may exist in a particular case, and the profound effect such advice may have on the social as well as the sexual life of parents. Genetic counselling should therefore never be given lightly.

References

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Conference Report

British Medical Association

Annual Scientific Meeting, 1975

The Annual Scientific Meeting of the British Medical Association was held at the University of Leeds between 7 and 11 July. It consisted of a series of clinic visits and two scientific symposia; one on 9 July a Ciba symposium on Primary Care and the Elderly, the other on 10 July a Boots symposium on Arthritis and its Treatment.

Primary Care and the Elderly

The Chairman, Professor Sir Ferguson Anderson (Glasgow), welcomed the speakers and indicated that the choice of topic, "Primary Care of the Elderly," was particularly appropriate at a time when projections for health care needs in the next 25 years indicated an expansion of 122% of the elderly over 85 years of age. He outlined the desirability of a mandatory period of training within a geriatric unit for all medical students before qualification, and suggested that such a training period could be extended as the proper remit of physicians, orthopaedic surgeons, and even anaesthetists.

Requirements of the Elderly

Mr. David Hobman (Director, Age Concern, England) began by emphasizing that the needs of the elderly differed in no way from his own personal requirements. In particular, the choice of lifestyle, the need for adequate income, and the desire for reasonable comfort, warmth, and independence in familiar home surroundings were all regarded as essential. Access to information on the social services available to the elderly was required if further hardship was to be prevented.

Mr. Hobman continued the theme of individual independence saying that the elderly needed a telephone (less than 25% of the retired population has access to a telephone), adequate transport facilities, and a precooked home-meals service. In the event of residential care being necessary, the option to withdraw, and privacy within a community were fundamental rights for each elderly person. When terminal illness was presented the need for the practitioner as friend and comforter was emphasized.

Though the doctor was central in the primary care team, he must learn more about the needs of the elderly, by varying and increasing routes of access to him by those most in need. Further use of the social service agencies providing care was required on a team basis, and this itself could lead to breakdown of barriers between health agencies and the education of medical students in the provision of better care for the elderly in the future—a reform which Mr. Hobman saw as solving the problem of our time.

Objectives in Geriatrics

Professor James Williamson (Liverpool) then outlined the need for care of the elderly, the responsibilities of the profession in providing that requirement, and the value of a management scheme designed to provide optimal benefits with available resources. He accepted that much progress had been made in the past 20 years with the establishment of over 200 departments of geriatric medicine in N.H.S. hospitals (with six chairs), and the agreement of geriatricians on the basic requirements for an effective service.

The difficulty of allocating resources within the specialty was highlighted by at least 40 unfilled consultant geriatrician vacancies, a trend to emigration from doctors of consultant